DISEASE:
Autosomal recessive multiple pterygium syndrome

NAME:	Autosomal recessive multiple pterygium syndrome
DESCRIPTION:	A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by congenital pterygia (webbing) mainly affecting the neck and large joints, arthrogryposis multiplex, short stature, and craniofacial dysmorphism (including ptosis, downslanting palpebral fissures, high-arched palate, and retrognathia). Additional manifestations are decreased movements, facial weakness, respiratory distress, vertebral anomalies, scoliosis, anomalies of the fingers, and cryptorchidism, among others. The disease is a non-lethal variant of multiple pterygium syndrome.
ORPHACODE:	2990
SYNONYMS:	Autosomal recessive non-lethal multiple pterygium syndrome EVMPS Escobar syndrome Escobar variant multiple pterygium syndrome
XREF(S):	Orphanet OMIM ICD-10 OMIM
ANALYTE(S):	CHRNG MYH3

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